

Applicants: Nathan Ellis, James German, and Joanna Groden  
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page: 11

Amendments to the Claims:

Please cancel Claims 13 and 18 without disclaimer or prejudice to applicants' right to pursue the subject matter of these claims in a future continuation or divisional application. Please amend Claim 11 and add new Claims 86-93 as set forth below.

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1-10. (Canceled)

11. (Currently Amended) A method for determining whether a subject is a carrier of a mutated *BLM* gene comprising detecting the presence of a mutated *BLM* gene in nucleic acid of said subject, wherein the mutated *BLM* gene is a mutated form of nucleic acid encoding the amino acid sequence set forth in SEQ ID NO:78.

12. (Original) The method of Claim 11 wherein the subject is an embryo, fetus, newborn, infant or adult.

E13  
13. (Canceled)

14. (Original) The method of Claim 11 wherein the nucleic acid is DNA or RNA.

15. (Original) The method of Claim 11 wherein the presence of the mutated *BLM* gene is detected by one or more techniques selected from the group consisting of sequence analysis, restriction enzyme digestion analysis, hybridization and polymerase chain reaction.

16. (Original) The method of Claim 11 wherein the presence of the mutated *BLM* gene is detected by the presence of a gene product encoded by the mutated *BLM* gene.

Applicants: Nathan Ellis, James German, and Joanna Groden  
Serial No.: 09/753,143  
Filed: January 2, 2001  
page: 12

17. (Original) The method of Claim 16 wherein the gene product is a protein.

18. (Canceled)

19. (Original) The method of Claim 16 wherein the gene product is mRNA.

20. (Original) The method of Claim 19 wherein the mRNA is detected by one or more techniques selected from the group consisting of sequence analysis, hybridization and polymerase chain reaction.

21-85. (Canceled)

E<sup>13</sup>  
86. (New) The method of Claim 11, wherein the mutated *BLM* gene is characterized by one or more of: (i) a deletion of nucleotides 631-633 of SEQ ID NO:72; (ii) a substitution of A with T at nucleotide 888 of SEQ ID NO:72; (iii) an insertion of A after nucleotide 1610 of SEQ ID NO:72; (iv) a substitution of A with G at nucleotide 2089 of SEQ ID NO:72; (v) a replacement of nucleotides ATCTGA at position 2281-2286 of SEQ ID NO:72 with nucleotides TAGATTC; (vi) a substitution of T with C at nucleotide 2596 of SEQ ID NO:72; and (vii) a substitution of G with C at nucleotide 3238 of SEQ ID NO:72.

87. (New) The method of Claim 86, wherein the mutated *BLM* gene is characterized by a deletion of nucleotides 631-633 of SEQ ID NO:72.

88. (New) The method of Claim 86, wherein the mutated *BLM* gene is

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Serial No.: 09/753,143  
Filed: January 2, 2001  
page: 13

characterized by a substitution of A with T at nucleotide 888 of SEQ ID NO:72.

89. (New) The method of Claim 86, wherein the mutated *BLM* gene is characterized by an insertion of A after nucleotide 1610 of SEQ ID NO:72.

90. (New) The method of Claim 86, wherein the mutated *BLM* gene is characterized by a substitution of A with G at nucleotide 2089 of SEQ ID NO:72.

91. (New) The method of Claim 86, wherein the mutated *BLM* gene is characterized by a replacement of nucleotides ATCTGA at position 2281-2286 of SEQ ID NO:72 with nucleotides TAGATTG.  
*E 13*

92. (New) The method of Claim 86, wherein the mutated *BLM* gene is characterized by a substitution of T with C at nucleotide 2596 of SEQ ID NO:72.

93. (New) The method of Claim 86, wherein the mutated *BLM* gene is characterized by a substitution of G with C at nucleotide 3238 of SEQ ID NO:72.

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